Case Report

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Eighteen Years with Spinal Muscular Atrophy (SMA) Type 1

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INTRODUCTION

SMA-1, also known as Werdnig-Hoffmann disease, is the most common neuromuscular disease of hypotonic newborns. Its incidence is over 1 per 8000 births and is a common cause of sudden infant death syndrome. It is characterized by the degeneration of the anterior horn cells of the spinal cord and of the motor nuclei in the lower brainstem (1). About 99% of patients have deletions in exons 6 and 7 of chromosome 5q13 causing a decrease in the transcription of survival motor neuron (SMN) protein. The SMAs are classified as types 1 through 4. Children with type 1 are hypotonic at birth, never sit independently, and 85% develop acute respiratory failure (ARF) before 18 months of age and the remainder, subsequently (2). Patients with severe SMA-1 either die or become continuously respirator-dependent before 6 months of age; patients with typical SMA-1 develop ARF during otherwise benign upper respiratory tract infections (URIs) at 6 to 18 months of age; and the 10 to 15% with mild SMA-1 develop ARF during URIs (2). When intubated for acute ARF, only 6% of the conventionally managed typical SMA-1 patients are successfully extubated without undergoing tracheotomy (3).

Although there are numerous reports of severe and typical SMA-1 patients dying from ARF before 18 months of age (4-6), it has also been reported that these patients can survive for years when undergoing tracheotomy for long-term tracheostomy mechanical ventilation (TMV) (3,7). Thus far, however, only one center has reported prolonged survival in SMA-1 by continuous long-term noninvasive intermittent positive pressure ventilatory support (CNVS) or high span (inspiratory positive airway pressure [IPAP], expiratory PAP [EPAP] spans greater than 10 cm H2O) bi-level PAP without the need to resort to tracheotomy (3). This has led to intubated children with SMA-1 being flown to that center from their native lands to avoid tracheotomy by being successfully extubated despite having failed extubation attempts and all spontaneous breathing trials (8). In some cases, families have emigrated to avoid their children from ever having to undergo tracheotomy. This is a report of one such patient who, along with her family, emigrated from Iran to prolong her survival without undergoing tracheotomy.

CASE SUMMARIES

A girl born in Tehran in August of 1994 was hypotonic at birth and noted to have progressive weakening by 3

months of age at which point she was diagnosed with SMA-1 and she and her family immigrated to the United States. She first developed a URI that resulted in a 3-month stay in a local hospital for pneumonia and ARF (URI-pneumonia) due principally to a weak cough at 8 months of age for which she was intubated and had a gastrostomy tube placed for all future nutrition. After three failed extubation attempts to low span bi-level PAP at IPAP 8 cm H2O and EPAP 4 cm H2O, no back-up rate, and receiving supplemental oxygen, a clinician experienced in extubating patients who fail all spontaneous breathing attempts (author JRB) attended the fourth attempt and successfully extubated her to high span bi-level PAP with IPAP 16, EPAP 4, rate 16 per minute. She was discharged home using nocturnal high span nasal bi-level PAP which she began to require for daytime as well as nocturnal support from age 2 and continues to use with the same settings.

From age 1 until age 8 she had 7 additional URI-pneumonias for which she was hospitalized locally and intubated. On three of those occasions she was successfully extubated to high span bi-level PAP on the first attempt and on four occasions on the second attempt for a total of 11 intubations. Mechanically assisted coughing (MAC), or mechanical insufflation-exsufflation along with an exsufflation-timed abdominal thrust (CoughAssist™, Philips-Respironics International Inc., Murraysville, Pa) were not used (9).

In 2003, at 9 years of age, with a vital capacity (VC) of 130 ml (7% of normal) (10,11), she was again intubated locally for a URI-pneumonia but failed 3 extubation attempts to high span bi-level PAP because of difficulty clearing airway secretions, and was told to undergo tracheotomy. At this point she was transferred to the second author's service where MAC was used aggressively through the translaryngeal tube to normalize SpO2 without supplemental oxygen, and then despite having no measurable VC prior to extubation, she was extubated successfully to high span bi-level PAP and aggressive MAC. The patient and her family were trained in MAC and administered it every 30 to 60 minutes post-extubation to keep, or return oxyhemoglobin saturation (SpO2) levels over 94% as described for other conditions (12). She was discharged with home access to MAC to avoid future URI-pneumonias.

From 2003 to the present time she has been using MAC during yearly URIs to maintain her baseline SpO2 at 95% or greater without supplemental oxygen, and, thereby, required only one hospitalization and intubation and was again successfully extubated to high span bi-level PAP and MAC despite having a VC of 30 ml prior to and 70 ml immediately after extubation. Post-extubation, the MAC was applied by the family every 30 to 60 minutes while she was awake. She also had three additional successful extubations to high span bi-level PAP and MAC following general anesthesia for surgical procedures. The family and the center's physicians attribute the avoidance of annual intubations, elimination of any extubation failures, and avoidance of tracheotomy over the last 10 years to the effective use of MAC as well as to high span bi-level PAP to maintain normal SpO2, especially during URIs.

Table 1 denotes the patient's VCs over time. The maximum she ever had was 170 ml or 5% of predicted normal (10,11). Currently, at age 18, she has a VC of 120 ml (2% of normal), no measurable cough flows, and requires high span bi-level PAP continuously via nasal interfaces (Figure 1) with less than 2 minute periods free from it before respiratory distress and blood gas derangement necessitate return to ventilatory support. She continues to have no volitional movement below her neck but has some eye, eye lid, forehead movement, and can wiggle her ears to operate a computer cursor for computer generated speech. She lives with her parents and has never had any other support in the home. She recently attended the wedding of her sister, has a Facebook account with which she communicates with her friends, and as her mother asserts, "she loves the life she has".

Table 1. The Patients' VC Over Time.

Age (years)	Vital Capacity (ml)	Vital Capacity % of Predicted Normal
5.3	75	7
5.9	100	7
6.8	110	7
8.2	150	8
9.4	150	7
11.0	170	6
12.7	170	5
14.0	120	4
17.9	115	2

*Immediately preceding extubation

** Within 24 hour post-extubation

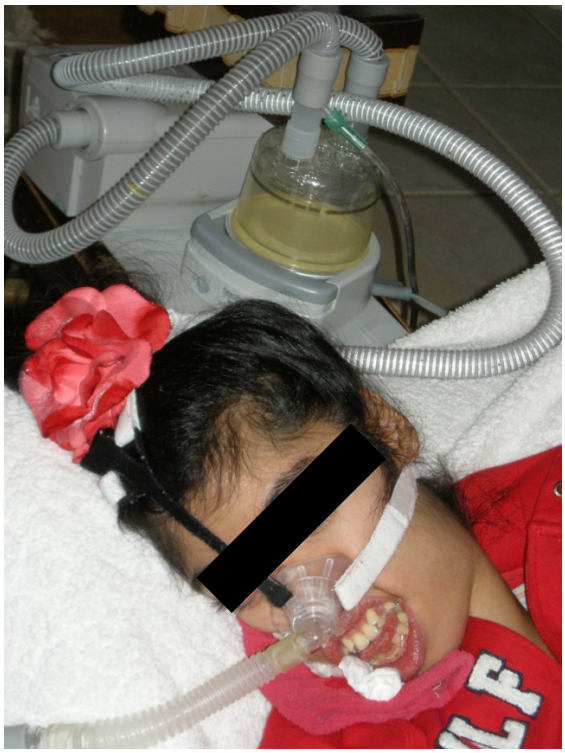

Figure 1. Patient with typical spinal muscular atrophy type 1 using continuous noninvasive ventilatory support via nasal interface.

DISCUSSION

Conventional extubation attempts are done for patients receiving supplemental oxygen, thereby depressing ventilatory drive, exacerbating hypercapnia, and rendering the oximeter useless as a gauge of alveolar ventilation and airway congestion (13) and without noninvasive ventilatory support or MAC. A protocol specifically designed for extubating SMA-1 patients who can not pass SB trials either before or after extubation was reported in 2000 (8), and used for this patient multiple times when ventilator weaning was impossible. The same protocol was used to successfully extubate 155 out of 157 unweanable older children and adults with SMA and other conditions in 2010 (14), as well as to extubate unweanable infants with SMA-1 (3). Both portable ventilators, and bi-level PAP units when used at high spans, can provide full ventilatory support even to patients with no measurable VC and can be used indefinitely as an alternative to TMV (14).

Although mechanical insufflator-exsufflators are unavailable in Iran, MAC is critical to prevent morbidity and mortality in patients with ineffective cough flows. As an alternative, patients have used manual resuscitators to "air stack" to deep lung volumes (15), then their vacuum cleaners via mouth pieces or other facial interfaces along with exsufflation-timed abdominal thrusts. These methods along with MAC have the potential to greatly prolong the lives of patients with neuromuscular disease while avoiding hospitalizations, tracheostomy tubes, and episodes of ARF. Mechanical insufflator-exsufflators are also used to deliver deep insufflations multiple times daily for lung volume recruitment that can promote lung growth, help maintain pulmonary compliance, limit pulmonary restriction, prevent pectus excavatum (16), and, thereby, further enhance the ability to generate effective cough flows. Patients managed noninvasively also better retain the ability to speak and some freedom from continuous ventilatory support than patients using TMV and they avoid death from tracheostomy tube associated complications (17). Also, all reported TMV patients who have been decanulated and switched to noninvasive management have preferred the latter for comfort, ability to speak, swallow, appearance, safety, and overall (18).

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